

dbGaP Study Release Notes



Release Notes for NHLBI TOPMed WGS VUH AF, phs001032.v6.p2

"NHLBI TOPMed: The Vanderbilt Atrial Fibrillation Registry"

For any questions or comments, please contact: dbgap-help@ncbi.nlm.nih.gov.

| | | |
|----------|----------|---------------------------------|
| October | 19, 2016 | Version 1 Data set release date |
| January | 25, 2017 | Version 2 Data set release date |
| December | 13, 2017 | Version 3 Data set release date |
| January | 9, 2019 | Version 4 Data set release date |
| January | 24, 2020 | Version 5 Data set release date |
| June | 3, 2021 | Version 6 Data set release date |

2021-06-03

Version 6 Data set release for NHLBI TOPMed WGS VUH AF now available

This release includes the addition of Freeze 9 whole genome sequences (WGS) and corresponding VCFs. Please refer to the latest study configuration report for a detailed description of each download component.

Consent group 1 (c1): General Research Use (IRB) (GRU-IRB)

| Data Type | subjects | samples |
|------------------------|----------|---------|
| Phenotype | 1134 | 1134 |
| Seq_DNA_SNP_CNV (VCFs) | 1122 | 1122 |
| WGS* | 1122 | 1122 |

*These data are brokered through the Sequence Read Archive (SRA). Please see Authorized Access instructions below.

For a description of non-SRA SAMPLE_USE terms, please see:

<https://www.ncbi.nlm.nih.gov/projects/gap/submission/GetSampleUseTypes.cgi>

Study and Phenotype Data Updates

1. New Study Accession

NHLBI TOPMed WGS VUH AF version 5 phs001032.v5.p2 has been updated to version 6. The dbGaP accession for the current set of data is **phs001032.v6.p2**. The participant number (p#) has not changed in version 6. No new subjects have been added to this study.

2. There are no updates to the phenotype datasets.

Molecular Data Updates

Two genomic accessions, phg001326.v1 freeze 8 and phg001566.v1 freeze9, are associated with the study.

1. See download components 'sample-info' for manifest of genotyped samples and files.
2. Genotype calls are available in matrix format ('genotype-calls-vcf') as VCF file(s) with samples of same data sharing consent.
3. Quality control data are in 'genotype-qc' tars.

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| phg | freeze | sample_cnt | subject_cnt |
|--------------|--------|------------|-------------|
| phg001326.v1 | 8 | 1110 | 1110 |
| phg001566.v1 | 9 | 1122 | 1122 |

Authorized Access (Individual Level Data and SRA Data)

Individual level data and Sequence Read Archive (SRA) data are available for download through the dbGaP Authorized Access System upon approval of the Data Access Request (DAR):

- <https://dbgap.ncbi.nlm.nih.gov/aa/wga.cgi?login=&page=login>

Public FTP site (Summary Level Data Only)

All data tables, data dictionaries, and documents will be housed under one directory for ease of downloading. The data_dict filenames have an added study version number (phs#.v#) and deleted participant set number (p#) from the table accession (pht#.v#). The var_report filenames have an added study version number (phs#.v#). In the var_report files, variables contain version numbers (phv#.v#) and summaries were created for each consent group (c#). These FTP files are available at:

- <https://ftp.ncbi.nlm.nih.gov/dbgap/studies/phs001032/phs001032.v6.p2>

2020-01-24

Version 5 Data set release for NHLBI TOPMed WGS VUH AF now available

This release includes the addition of Freeze 8 whole genome sequences (WGS) brokered through the Sequence Read Archive (SRA), and VCFs derived from WGS. Please refer to the latest study configuration report for a detailed description of each download component.

Consent group 1 (c1): General Research Use (IRB) (GRU-IRB)

| Data Type | subjects | samples |
|------------------------|----------|---------|
| Phenotype | 1134 | 1134 |
| Seq_DNA_SNP_CNV (VCFs) | 1110 | 1110 |
| WGS* | 1110 | 1110 |

*These data are brokered through the Sequence Read Archive (SRA). Please see Authorized Access instructions below.

For a description of non-SRA SAMPLE_USE terms, please see:

<https://www.ncbi.nlm.nih.gov/projects/gap/submission/GetSampleUseTypes.cgi>

Study and Phenotype Data Updates

1. New Study Accession

NHLBI TOPMed WGS VUH AF version 4 phs001032.v4.p2 has been updated to version 5. The dbGaP accession for the current set of data is **phs001032.v5.p2**. The participant number (p#) has not changed in version 5. No new subjects have been added to this study.

2. There are no updated datasets.

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Molecular Data Updates

1. See download components phg001326.v1.TOPMed_WGS_Vanderbilt_AF_v5.sample-info.MULTI.tar for manifest of genotyped samples and files.
2. Genotype calls are available in matrix format ('genotype-calls-vcf') as VCF file(s) with samples of same data sharing consent.
3. Quality control data are in phg001326.v1.TOPMed_WGS_Vanderbilt_AF_v5.genotype-qc.MULTI.tar
4. Only Freeze 5b and Freeze 8 VCFs will be available for download.

| phg_name | sample_cnt | subject_cnt |
|--------------|------------|-------------|
| phg001138.v1 | 1018 | 1018 |
| phg001326.v1 | 1110 | 1110 |

Authorized Access (Individual Level Data and SRA Data)

Individual level data and Sequence Read Archive (SRA) data are available for download through the dbGaP Authorized Access System upon approval of the Data Access Request (DAR):

- <https://dbgap.ncbi.nlm.nih.gov/aa/wga.cgi?login=&page=login>

Public FTP site (Summary Level Data Only)

All data tables, data dictionaries, and documents will be housed under one directory for ease of downloading. The data_dict filenames have an added study version number (phs#.v#) and deleted participant set number (p#) from the table accession (pht#.v#). The var_report filenames have an added study version number (phs#.v#). In the var_report files, variables contain version numbers (phv#.v#) and summaries were created for each consent group (c#). These FTP files are available at:

- <https://ftp.ncbi.nlm.nih.gov/dbgap/studies/phs001032/phs001032.v5.p2>

2019-01-09

Version 4 Data set release for NHLBI TOPMed WGS VUH AF now available

This release includes updated phenotype tables, whole genome sequences (WGS) brokered through the SRA, and VCFs derived from WGS. Please refer to the latest study configuration report for a detailed description of each download component.

Consent group 1 (c1): General Research Use (IRB) (GRU-IRB)

| | Phenotype | Seq_DNA_SNP_CNV (VCFs) | WGS |
|----------|-----------|------------------------|------|
| subjects | 1134 | 1018 | 1134 |
| samples | 1134 | 1018 | 1134 |

Study and Phenotype Data Updates

1. New Study Accession

NHLBI TOPMed WGS VUH AF version 3 phs001032.v3.p2 has been updated to Version 4. The

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dbGaP accession for the current set of data is **phs001032.v4.p2**. The participant number (p#) has not changed in version 4. No new subjects have been added to this study.

2. Updated Datasets (n=1)

| pht | version | Dataset Name |
|------|---------|--------------------------|
| 5099 | 3 | TOPMed_WGS_VUH_AF_Sample |

- Please note we are discontinuing the submission and distribution of the SAMPLE_USE variable. The sample use counts will be populated by SRA (sequences) and dbGaP (all other submitted molecular data).

Molecular Data Updates

New genomics accession, phg001138 with genotypes on GRC38 is added to the study.

- See download components phg001138.v1.TOPMed_WGS_Vanderbilt_AF_v4.sample-info.MULTI.tar for manifest of genotyped samples and files.
- Genotype calls are available in matrix format ('genotype-calls-vcf') as VCF file(s) with samples of same data sharing consent.

| phg_name | sample_cnt | subject_cnt |
|--------------|------------|-------------|
| phg000991.v1 | 1110 | 1110 |
| phg001138.v1 | 1018 | 1018 |

Authorized Access (Individual Level Data and SRA Data)

Individual level data and Sequence Read Archive (SRA) data are available for download through the dbGaP Authorized Access System upon approval of the Data Access Request (DAR):

- <https://dbgap.ncbi.nlm.nih.gov/aa/wga.cgi?login=&page=login>

Public FTP site (Summary Level Data Only)

All data tables, data dictionaries, and documents will be housed under one directory for ease of downloading. The data_dict filenames have an added study version number (phs#.v#) and deleted participant set number (p#) from the table accession (pht#.v#). The var_report filenames have an added study version number (phs#.v#). In the var_report files, variables contain version numbers (phv#.v#) and summaries were created for each consent group (c#). These FTP files are available at:

- <https://ftp.ncbi.nlm.nih.gov/dbgap/studies/phs001032/phs001032.v4.p2>

2017-12-13

Version 3 Data set release for NHLBI TOPMed WGS VUH AF now available

This release includes updated phenotype tables, whole genome sequences (WGS) brokered through the SRA, and VCFs derived from WGS. Please refer to the latest study configuration report for a detailed description of each download component.

Consent group 1 (c1): General Research Use (IRB) (GRU-IRB)

| | Phenotype | Seq_DNA_SNP_CNV | Seq_DNA_WholeGenome |
|--|-----------|-----------------|---------------------|
|--|-----------|-----------------|---------------------|

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| | | | |
|----------|------|------|------|
| subjects | 1134 | 1110 | 1134 |
| samples | 1134 | 1110 | 1134 |

Molecular data descriptions:

(<https://www.ncbi.nlm.nih.gov/projects/gap/submission/GetSampleUseTypes.cgi>)

- Seq_DNA_SNP_CNV: SNP and CNV genotypes derived from sequence data (VCFs)
- Seq_DNA_WholeGenome: Whole genome sequencing

Study and Phenotype Data Updates

1. New Study Accession

NHLBI TOPMed WGS VUH AF version 2 phs001032.v2.p1 has been updated to Version 3. The dbGaP accession for the current set of data is **phs001032.v3.p2**. The participant number (p#) has changed in version 3; subjects have been retired. No new subjects have been added to this study.

2. New Datasets (n=1 dataset)

| pht | version | Dataset Name |
|------|---------|----------------------------|
| 7135 | 1 | TOPMed_WGS_VUH_AF_Pedigree |

3. Updated Datasets (n=4 datasets; all existing variables have been updated)

| pht | version | Dataset Name |
|------|---------|--------------------------------------|
| 5098 | 2 | TOPMed_WGS_VUH_AF_Subject |
| 5099 | 2 | TOPMed_WGS_VUH_AF_Sample |
| 5100 | 2 | TOPMed_WGS_VUH_AF_Sample_Attributes |
| 5675 | 3 | TOPMed_WGS_VUH_AF_Subject_Phenotypes |

4. New Variables (n=7 variables)

| pht | pht version | Dataset Name | phv | Variable Name |
|------|-------------|-------------------------------------|--------|-------------------|
| 5098 | 2 | TOPMed_WGS_VUH_AF_Subject | 328568 | SUBJECT_SOURCE |
| 5098 | 2 | TOPMed_WGS_VUH_AF_Subject | 328569 | SOURCE_SUBJECT_ID |
| 5100 | 2 | TOPMed_WGS_VUH_AF_Sample_Attributes | 328575 | SEQUENCING_CENTER |
| 5100 | 2 | TOPMed_WGS_VUH_AF_Sample_Attributes | 328576 | Funding_Source |
| 5100 | 2 | TOPMed_WGS_VUH_AF_Sample_Attributes | 328577 | TOPMed_Phase |
| 5100 | 2 | TOPMed_WGS_VUH_AF_Sample_Attributes | 328578 | TOPMed_Project |
| 5100 | 2 | TOPMed_WGS_VUH_AF_Sample_Attributes | 328579 | Study_Name |

Molecular Data Updates

- See download components phg000991.v1.TOPMed_WGS_Vanderbilt_AF_v3.sample-info.MULTI.tar and phg000991.v1.TOPMed_WGS_Vanderbilt_AF_v3.marker-info.MULTI.tar for manifest of genotyped samples and files, and information about marker set used for genotyping.
- Genotype calls are available in matrix format ('genotype-calls-vcf') as VCF file(s) with samples of same data sharing consent.
- Quality control data are in phg000991.v1.TOPMed_WGS_Vanderbilt_AF_v3.genotype-qc.MULTI.tar.

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Authorized Access (Individual Level Data and SRA Data)

Individual level data and Sequence Read Archive (SRA) data are available for download through the dbGaP Authorized Access System upon approval of the Data Access Request (DAR):

- <https://dbgap.ncbi.nlm.nih.gov/aa/wga.cgi?login=&page=login>

Public FTP site (Summary Level Data Only)

All data tables, data dictionaries, and documents will be housed under one directory for ease of downloading. The data_dict filenames have an added study version number (phs#.v#) and deleted participant set number (p#) from the table accession (pht#.v#). The var_report filenames have an added study version number (phs#.v#). In the var_report files, variables contain version numbers (phv#.v#) and summaries were created for each consent group (c#). These FTP files are available at:

- <https://ftp.ncbi.nlm.nih.gov/dbgap/studies/phs001032/phs001032.v3.p2>

2017-01-25

Version 2 Data set release for NHLBI TOPMed WGS VUH AF now available

This release includes changes to the subject phenotype data table. Please refer to the latest study configuration report for a detailed description of each download component.

Consent group 1 (c1): General Research Use (IRB) (GRU-IRB)

| | phenotype | SRA/VCFs |
|----------|-----------|----------|
| subjects | 1157 | 310 |
| samples | 1157 | 310 |

Study and Phenotype Data Updates

1. New Study Accession

NHLBI TOPMed WGS VUH AF version 1 phs001032.v1.p1 has been updated to Version 2. The dbGaP accession for the current set of data is **phs001032.v2.p1**. The participant number (p#) has not changed in version 2. No new subjects have been added to this study.

2. Deleted Variable (n=1)

| pht | Dataset Name | phv | Variable Name |
|------|--------------------------------------|--------|---------------|
| 5675 | TOPMed_WGS_VUH_AF_Subject_Phenotypes | 265820 | EF_ever_50 |

Authorized Access (Individual Level Data and SRA Data)

Individual level data and SRA sequencing data are available for download through the dbGaP Authorized Access System upon approval of the Data Access Request (DAR):

- <https://dbgap.ncbi.nlm.nih.gov/aa/wga.cgi?login=&page=login>

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Public FTP site (Summary Level Data Only)

All data tables, data dictionaries, and documents will be housed under one directory for ease of downloading. The data_dict filenames have an added study version number (phs#.v#) and deleted participant set number (p#) from the table accession (pht#.v#). The var_report filenames have an added study version number (phs#.v#). In the var_report files, variables contain version numbers (phv#.v#) and summaries were created for each consent group (c#). These FTP files are available at:

- <https://ftp.ncbi.nlm.nih.gov/dbgap/studies/phs001032/phs001032.v2.p1>

2016-10-19

Version 1 Data set release for NHLBI TOPMed WGS VUH AF now available

This release includes TOPMed Phase I phenotype tables, whole genome sequences (WGS) brokered through the SRA, and VCFs derived from WGS. Additionally, phenotype tables include subjects and samples beyond TOPMed Phase I in order to instantiate IDs for future versions. Please refer to the latest study configuration report for a detailed description of each download component.

Consent group 1 (c1): General Research Use (IRB) (GRU-IRB)

| | phenotype | SRA/VCFs |
|----------|-----------|----------|
| subjects | 1157 | 310 |
| samples | 1157 | 310 |

Molecular Data Updates

1. See download components phg000796.v1.TOPMed_WGS_Vanderbilt_AF.sample-info.MULTI.tar for manifest of genotyped samples.
2. Genotype calls are available in matrix format ('genotype-calls-vcf') as VCF file(s) with samples of same data sharing consent.
3. Quality control data are in phg000796.v1.TOPMed_WGS_Vanderbilt_AF.genotype-qc.MULTI.tar.

Authorized Access (Individual Level Data and SRA Data)

Individual level data and SRA sequencing data are available for download through the dbGaP Authorized Access System upon approval of the Data Access Request (DAR):

- <https://dbgap.ncbi.nlm.nih.gov/aa/wga.cgi?login=&page=login>

Public FTP site (Summary Level Data Only)

All data tables, data dictionaries, and documents will be housed under one directory for ease of downloading. The data_dict filenames have an added study version number (phs#.v#) and deleted participant set number (p#) from the table accession (pht#.v#). The var_report filenames have an added study version number (phs#.v#). In the var_report files, variables contain version numbers (phv#.v#) and summaries were created for each consent group (c#). These FTP files are available at:

- <https://ftp.ncbi.nlm.nih.gov/dbgap/studies/phs001032/phs001032.v1.p1>